

Supplemental Data

Homeotic Arm-to-Leg Transformation Associated with Genomic Rearrangements at the *PITX1* Locus

Malte Spielmann, Francesco Brancati, Peter Krawitz, Peter N. Robinson, Daniel Ibrahim, Martin Franke, Jochen Hecht, Silke Lohan, Katarina Däthe, Anna Maria Nardone, Paola Ferrari, Antonio Landi, Lars Wittler, Bernd Timmermann, Danny Chan, Ulrich Mennen, Eva Klopocki, and Stefan Mundlos

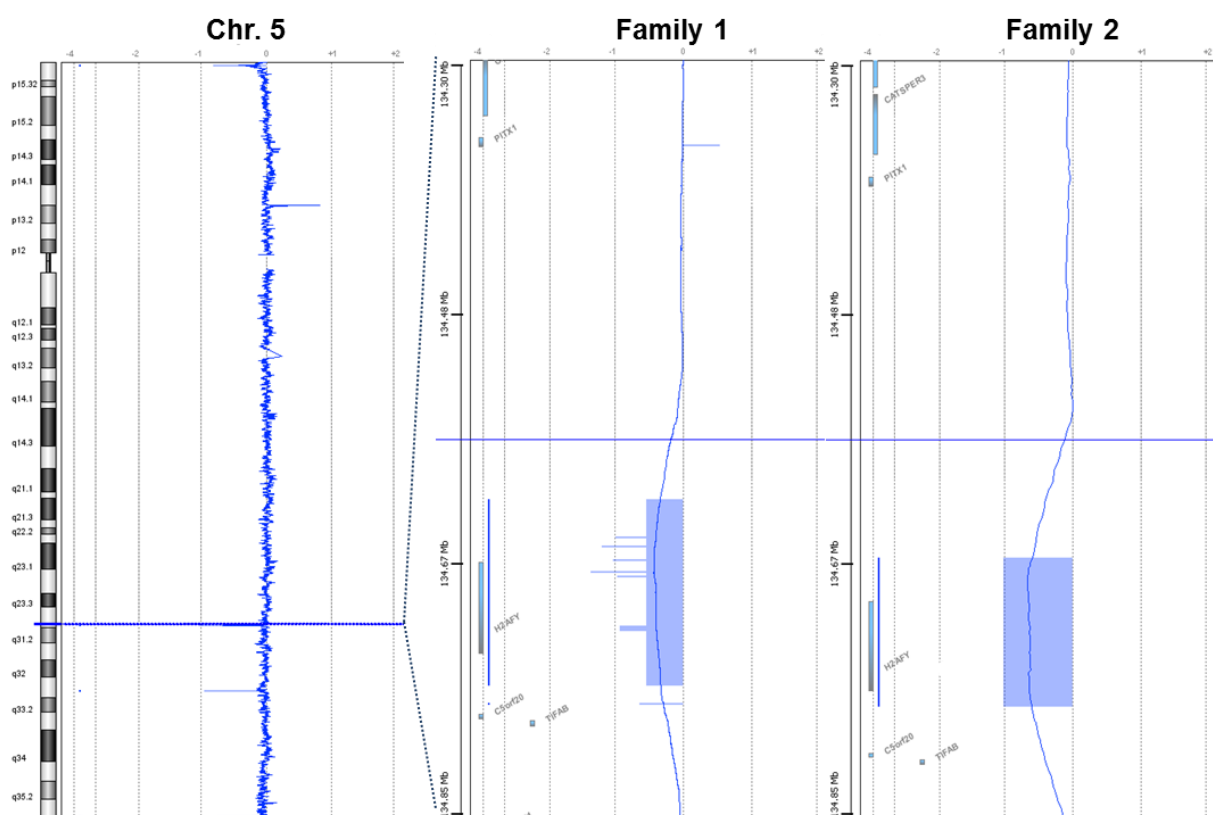


Figure S1. Array CGH Profile of Chromosome 5

On the left, whole chromosome 5; on the right details of the deleted regions in family 1 (chr5: 134,624,602–134,759,492 hg19) and family 2 (chr5: 134,638,524–134,746,407 hg19). The areas highlighted in blue indicate the extension of the heterozygous deletions. The deleted regions are overlapping and located 5' of *PITX1* separated by a gene desert.



Figure S2. A Translocation Breakpoint between chr5 and chr18 with a Resolution of 1 kb Was Detected by Screening for a Significant Enrichment of Translocated Reads

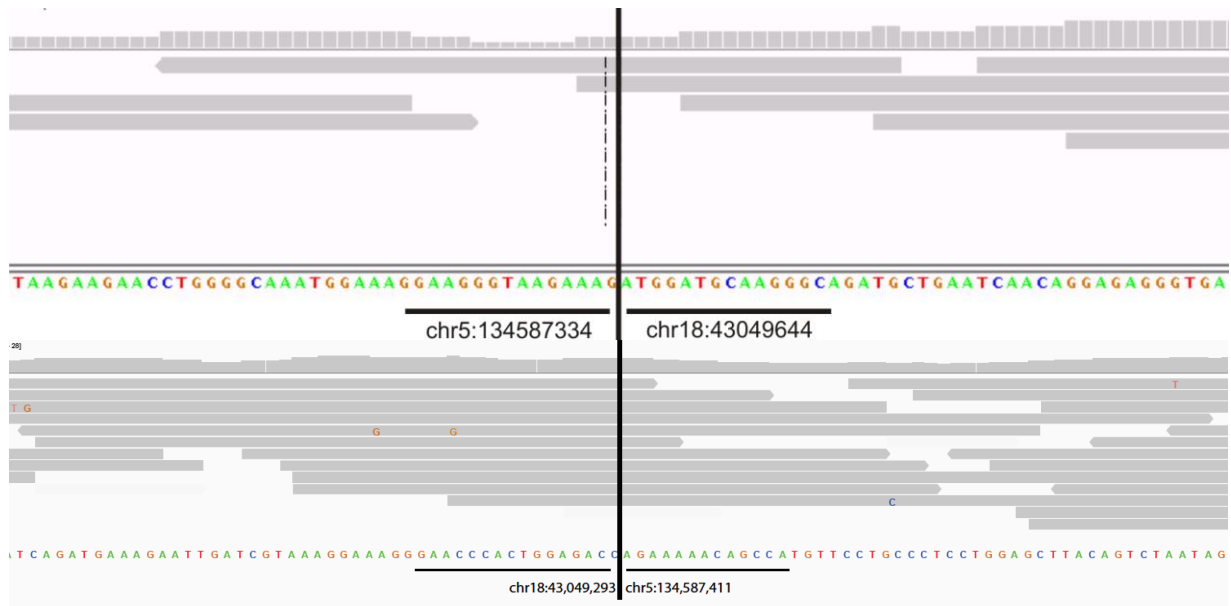


Figure S3. All Reads that Did Not Align to the Reference Sequence hg19 Were Mapped to Templates of Potential Breakpoints

IGV screenshot of two reads that map to a template on the derivative chromosome 5 at hg19 (chr5: 134,587,334; chr18: 430,49,644) and 11 reads that map to a template on the derivative chromosome 18 at hg19 (chr18: 43,049,293; chr5: 134,587,638).

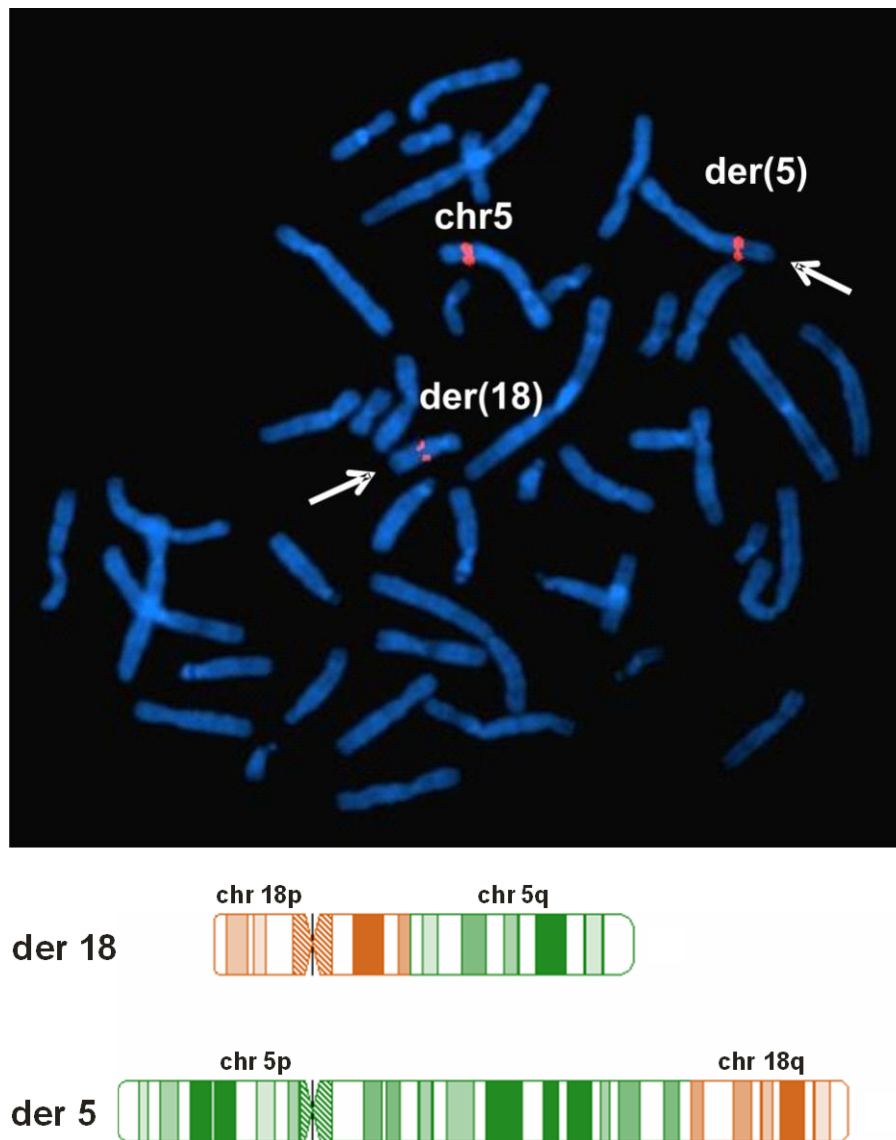


Figure S4. FISH Analysis on Metaphases from Cultured Lymphocytes Using the BAC Clone RP11-798G24 Mapping to 5q31.1 (chr5: 134,469,143–134,675,260 bp) Labeled with Spectrum Orange

Three signals are detected i.e. on the normal chromosome 5, on the derivative chromosome 5 and on the derivative chromosome 18 (arrows). The ideograms indicate the composition of the two derivate chromosomes resulting from this reciprocal translocation.